Aromatase excess syndrome

Description

Aromatase excess syndrome is a condition characterized by elevated levels of the female sex hormone estrogen in both males and females. Males with aromatase excess syndrome experience breast enlargement (gynecomastia) in late childhood or adolescence. The bones of affected males grow and develop more quickly and stop growing sooner than usual (advanced bone age). As a result males have an early growth spurt, typically during late childhood, with short stature as an adult. Affected females rarely show signs and symptoms of the condition, but they may have increased breast growth (macromastia), irregular menstrual periods, and short stature. The ability to have children (fertility) is usually normal in both males and females with aromatase excess syndrome.

Frequency

The prevalence of aromatase excess syndrome is unknown; more than 20 cases have been described in the medical literature.

Causes

Rearrangements of genetic material involving the *CYP19A1* gene cause aromatase excess syndrome. The *CYP19A1* gene provides instructions for making an enzyme called aromatase. This enzyme converts a class of hormones called androgens, which are involved in male sexual development, to different forms of estrogen. In females, estrogen guides female sexual development before birth and during puberty. In both males and females, estrogen plays a role in regulating bone growth.

The condition can result from several types of genetic rearrangements involving the *CYP19A1* gene. These rearrangements alter the activity of the gene and lead to an increase in aromatase production. In affected males, the increased aromatase and subsequent conversion of androgens to estrogen are responsible for the gynecomastia and limited bone growth characteristic of aromatase excess syndrome. Increased estrogen in females can cause symptoms such as irregular menstrual periods and short stature.

Learn more about the gene associated with Aromatase excess syndrome

- CYP19A1

Inheritance

This condition is inherited in an autosomal dominant pattern, which means a genetic rearrangement involving one copy of the *CYP19A1* gene in each cell is sufficient to cause the disorder.

In some cases, an affected person inherits the mutation from one affected parent. Other cases result from new genetic changes and occur in people with no history of the disorder in their family.

Other Names for This Condition

- AEXS
- Familial gynecomastia due to increased aromatase activity
- Hereditary gynecomastia
- Increased aromatase activity

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Aromatase excess syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1970109/)

Genetic and Rare Diseases Information Center

- Aromatase excess syndrome (https://rarediseases.info.nih.gov/diseases/12494/aromatase-excess-syndrome)

Patient Support and Advocacy Resources

- Disease InfoSearch (https://www.diseaseinfosearch.org/)
- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- AROMATASE EXCESS SYNDROME (https://omim.org/entry/139300)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28aromatase+excess+syndrome%5BTIAB%5D%29+OR+%28increased+aromatase+activity%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+day

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